Objective: Familial hypercholesterolemia (FH) is an autosomal dominant disease characterized with defective cholesterol removal from circulation and hypercholesterolemia caused by mutations in \textit{LDLR} gene. Although definitive diagnosis is made by mutation analysis, some diagnostic criteria have been developed due to the impracticality of routine mutation analysis. This study aims to compare mutation status of familial hypercholesterolemia patients and their diagnoses according to the Simone Broome criteria (SBC).

Material and Methods: All patients with primary hyperlipidemia followed up in the Metabolism and Nutrition Unit of Hacettepe University Medical School were classified according to the Frederickson classification system. Patients with type IIa hyperlipoproteinemia was assessed by SBC for the diagnosis of FH. Denaturing high-performance liquid chromatography (DHPLC) and DNA sequencing was performed on the most frequently mutated exons (9 and 12). Mutation analyses of this and previous cohorts were compared with SBC in terms of diagnostic compatibility.

Results: 118 of 163 patients with primary hyperlipidemia were type IIa hyperlipoproteinemia according to Frederickson classification. According to the SBC, 48% of these patients were in the “probable FH”, 26% were in “definite FH”, 17% were in “no FH” group, and 9% could not be classified due to insufficient information. On exon 9 heterozygous I420N(C.1322T>A) mutation was seen in 2 patients and on exon 12 and one heterozygous and one homozygous W556R(C.1729T>C) mutation were observed. When we combined these results with 29 patients results that were previously screened, mutations were seen in 10 out of 10 patients (100%) whose mutation statuses were analyzed in the definite FH, 12 out of 16 (75%) patients in the probable FH and 1 out of 3 (33%) patients in the no FH group.

Conclusion: When routine mutation screening is not a feasible choice or when selective screening is performed, diagnosing familial hypercholesterolemia based on the Simone Broome criteria is a suitable approach.